Rare Disesae Common Data Model (v2.0) based on ERDRI-CDS, HL7 FHIR® and GA4GH Phenopackets©

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Section	Data Element	Code	Terminology	Value Type	Value Property	Concepts			FHIR Expression	GA4GH Phenopacket element	Description
						Display Name	Code	Code System			
1. Formal Criteria	**1.1 Pseudonym**	422549004	SCT	Identifier	n/a	n/a			Patient.identifier	Individual.id	The (local) patient-related Identification code
	1.2 Date of Admission	399423000	SCT	Date	YYYY-MM-DD	n/a			Encounter.plannedStartDate	Individual. time_at_last_encounter	The date of admission or data capture of the individual
2. Personal Information	**2.1 Date of birth**	184099003	SCT	Date	YYYY-MM-DD	n/a			Patient.birthDate	Individual.date_of_birth	The individual's date of birth
	2.2 Sex at birth	281053000	SCT	Code	VSe	Female	248152002	SCT	Patient.extension:birth-sex Rec. VS: Recorded Sex Or Gender Type	Individual.sex	The individual's sex that was assigned at birth
						Male	248153007				
						Patient sex unknown	184115007				
						Intersex	32570691000036108				
						Not recorded	1220561009				
	2.3 Karyotypic Sex	karyotypic_sex	GA4GH	Code	VS	Unknown	UNKNOWN_KARYOTYPE	GA4GH	n/a	Individual.karyotypic_sex	The chromosomal sex of an individual
						XX	XX				
						XY	XY				
						XO	XO				
						XXY	XXY				
						XXYY	XXYY				
						XXXY	XXXY				
						XXXX	XXXX				
						XYY	XYY				
						Other	OTHER_KARYOTYPE				
	2.4 Gender	263495000	SCT	Code	VSe	Female	248152002	SCT	Patient.gender Recommended VS: AdministrativeGender	Individual.gender	The self-assigned gender of the individual
						Male	248153007				
						Gender unknown	394743007				
						Identifies as nonbinary gender	33791000087105				
						Not recorded	1220561009				
	2.5 Country of Birth	370159000	SCT	Address	VS	Country: use ISO 3166 - 2 or 3 letter country code			Patient.extension:patient-birthPlace	n/a	The individual's country of birth.
3. Vital Status	**3.1 Vital Status**	365860008	SCT	Code	VSe	Alive	438949009	SCT	(Patient.deceased.deceasedBoolean)	Individual.VitalStatus.status	The individual's general clinical status or vital status.
						Dead	419099009				
						Lost in follow-up	399307001				
						Opted-out	247755007				
						Unknown	261665006				
	3.2 Time of Death	398299004	SCT	Date	YYYY-MM-DD	n/a			Patient.deceased.deceasedDateTime	Individual.VitalStatus. time_of_death	If deceased, the individual's date of death.
	3.3 Cause of Death	16100001	SCT	Code	ICD-10	n/a			Observation.value.coding.code	Individual.VitalStatus. cause_of_death	If deceased, the individual's primary cause of death (i. e. according to the death certificate).

3. Patient Stati	***3.4 Age Category***	282032007	SCT	Code	VSe	Infancy	3658006	SCT	Patient.extension:age.value[x].coding.code	Individual.time_at_last_encounter.ontology_class	The individual's age category at the time of data capture (1.2).
						Toddler	713153009				
						Childhood	255398004				
						Adolescence	263659003				
						Adulthood	41847000				
						Fetal period	303112003				
						Dead	419099009				
						Unknown	261665006				
	3.5 Length of Gestation at Birth [weeks+days]	LP19507-0	LOINC	String	XX+X	n/a			n/a	n/a	The duration of the pregnancy in weeks and days, from the first day of the last menstrual period to the day of delivery, formatted as XX+X (weeks+days).
	3.6 Undiagnosed RD Case	*103330002*	SCT	Boolean	VSe	Yes	373066001	SCT	n/a	n/a	Identifies cases where an RD diagnosis has not been established.
						No	373067005				
4. Care Pathway	**4.1 Encounter Start**	encounter.period.start	HL7 FHIR	Date	YYYY-MM-DD	n/a			Encounter.period.start	n/a	The beginning of an encounter of the individual.
	4.2 Encounter End	encounter.period.end	HL7 FHIR	Date	YYYY-MM-DD	n/a			Encounter.period.end	n/a	The end of an encounter of the individual.
	4.3 Encounter Status	305058001	SCT	Code	VS	Planned	planned	HL7 FHIR	Encounter.status	n/a	The status of an encounter of the individual at the time of data capture.
						Arrived	arrived				
						Triaged	triaged				
						In Progress	in-progress				
						On Leave	onleave				
						Finished	finished				
						Cancelled	cancelled				
						Entered in Error	entered-in-error				
						Unknown	unknown				
	4.4 Encounter Class	encounter.class	HL7 FHIR	Code	VSe	Ambulatory	AMB	HL7 FHIR	Encounter.class Recommended VS: Encounter.class	n/a	The class of an encounter of the individual at the time of data capture.
						Inpatient	IMP				
						Observation	OBSENC				
						Emergency	EMER				
						Virtual	VR				
						Home Health	HH				
						RD Specialist Center	RDC	Own Encoding			
						Unknown	261665006	SCT			
	5.1 Disease	64572001	SCT	Code	ORDO	n/a			Condition.code	Disease.term: OntologyClass	A disease that the individual was affected by. If a genetic diagnosis or subtypes were diagnosed, please also provide the respective OMIM_g and OMIM_p codes.
					ICD-10						
					ICD-11						
					MONDO						
					OMIM_g						
					OMIM_p						
	5.2 Verification Status	99498-8	LOINC	Code	VS	Unconfirmed	unconfirmed	HL7 FHIR	Condition.verificationStatus	(Disease.excluded)	The verification status of the disease.
						Provisional	provisional				
						Differential	differential				
						Confirmed	confirmed				

5. Disease						Refuted	refuted				
						Entered in Error	entered-in-error				
	5.3 Age at Onset	424850005	SCT	Code	VSe	Prenatal	118189007	SCT	Condition.onsetPeriod	Disease.onset	The age at the onset of the first symptoms or signs of the disease.
						Birth	3950001				
						Date	410672004				
						Unknown	261665006				
	5.4 Date of Onset	298059007	SCT	Date	YYYY-MM-DD	n/a			Condition.onsetDateTime	Disease.onset	The date at onset of first symptoms or signs of the disease.
	5.5 Age at Diagnosis	423493009	SCT	Code	VSe	Prenatal	118189007	SCT	Condition.recordedDate	n/a	The individual's age when the diagnosis was made.
						Birth	3950001				
						Date	410672004				
						Unknown	261665006				
	5.6 Date of Diagnosis	432213005	SCT	Date	YYYY-MM-DD	n/a			Condition.recordedDate	Disease.onset	The date on which the disease was determined.
	5.7 Body Site	64572001: 363698007=442083009	SCT	Code	VS	[Include] descendent-of Code 123037004 Body structure (body structure)		SCT	Condition.bodySite.coding:snomed-ct	Disease.primary_site. OntologyClass	The specific body site affected by disease is encoded using all descendants of SCT Body Structure (123037004).
	5.8 Clinical Status	263493007	SCT	Code	VS	Active	active	HL7 FHIR	Condition.clinicalStatus	Interpretation. progress_status. ProgressStatus	The clinical status of the disease indicates whether it is active, inactive, or resolved.
						Recurrence	recurrence				
						Relapse	relapse				
						Inactive	inactive				
						Remission	remission				
						Resolved	resolved				
	5.9 Severity	64572001: 246112005=272141005	*SCT*	*Code*	VS	Severe	24484000	SCT	Condition.severity	n/a	The severity of the disease is categorised by clinical evaluation.
						Moderate	6736007				
						Mild	255604002				
	6.1.1 Genomic Diagnosis	106221001	SCT	Code	OMIM_p	n/a			Condition.code	Interpretation.Diagnosis. disease	The genomic diagnoses can correspond to the diagnosed disease in (5.1) if the same OMIM codes are used.
					OMIM_g						
	6.1.2 Progress Status of Interpretation	progress_status	GA4GH	Code	VS	No information is available about the diagnosis	UNKNOWN_PROGRESS	GA4GH	n/a	Interpretation. progress_status	The interpretation has a ProgressStatus that refers to the status of the attempted diagnosis.
						No diagnosis has been found to date but additional differential diagnostic work is in progress.	IN_PROGRESS				
						The work on the interpretation is complete.	COMPLETED				
						The interpretation is complete and also considered to be a definitive diagnosis	SOLVED				
						The interpretation is complete but no definitive diagnosis was found	UNSOLVED				
						No information is available about the status	UNKNOWN_STATUS				

	6.1.3 Interpretation Status	interpretation_status	GA4GH	Code	VS	The variant or gene reported here is interpreted not to be related to the diagnosis	REJECTED	GA4GH	n/a	Interpretation.GenomicInterpretation	An enumeration that describes the conclusion made about the genomic interpretation.
						The variant or gene reported here is interpreted to possibly be related to the diagnosis	CANDIDATE				
						The variant or gene reported here is interpreted to be related to the diagnosis	CONTRIBUTORY				
						The variant or gene reported here is interpreted to be causative of the diagnosis	CAUSATIVE				
	6.1.4 Structural Variant Analysis Method	LL4048-6	LOINC	Code	VS	Karyotyping	LA26406-1	LOINC	Observation.method	n/a	The method used to analyse structural variants in the genome.
						FISH	LA26404-6				
						PCR	LA26418-6				
						qPCR (real-time PCR)	LA26419-4				
						SNP array	LA26400-4				
						Restriction fragment length polymorphism (RFLP)	LA26813-8				
						DNA hybridization	LA26810-4				
						Sequencing	LA26398-0				
						MLPA	LA26415-2				
						Other	LA46-8				
	6.1.5 Reference Genome	62374-4	LOINC	Code	VS	NCBI Build 34 (hg16)	LA14032-9	LOINC	Observation.component:reference-sequence-assembly	[...].VariantInterpretation.VariationDescriptor.vrs_ref_allele_seq	The reference genome used for analysing the genetic variant.
						GRCh37 (hg19)	LA14029-5				
						NCBI Build 36.1 (hg18)	LA14030-3				
						NCBI Build 35 (hg17)	LA14031-1				
						GRCh38 (hg38)	LA26806-2				
	6.1.6 Genetic Mutation String	LP410543-5	LOINC	String	n/a	n/a			Observation.component:coding-hgvs.extension	[...]VariantInterpretation.VariationDescriptor.Extension	An unvalidated (HGVS) string that describes the variant change.
	6.1.7 Genomic DNA Change	81290-9	LOINC	Code	g.HGVS	n/a			Observation.component:genomic-hgvs.code	[...]VariantInterpretation.VariationDescriptor.Expression [+] [...].MoleculeContext	The specific change in the genomic DNA sequence encoded with a validated g.HGVS expression.
	6.1.8 Sequence DNA Change	48004-6	LOINC	Code	c.HGVS	n/a			Observation.component:coding-hgvs.code		The specific change in the DNA sequence at the nucleotide level with a validated c.HGVS expression
	6.1.9 Amino Acid Change	48005-3	LOINC	Code	p.HGVS	n/a			Observation.component:protein-hgvs.code		The specific change in the amino acid sequence resulting from a genetic variant as a validated p.HGVS expression
	6.1.10 Gene	48018-6	LOINC	Code	HGNC	n/a			Observation.component:gene-studied.code	[...].GenomicInterpretation.GeneDescriptor.value_id	The specific gene or genes that were analysed or identified in the study.
						Homozygous	LA6705-3		Observation.component:allelic-state	[...].GenomicInterpretation.VariantInterpretation	
						(simple) Heterozygous	LA6706-1				
						Compound heterozygous	LA26217-2				

6.1 Genetic Findings

	6.1.11 Zygosity	48007-9	LOINC	Code	VSe	Double heterozygous	LA26220-6	LOINC	Observation.component:allelic-state Rec. VS: AllelicState	variantInterpretation. variationDescriptor. AllelicState	The zygosity of the genetic variant.
						Hemizygous	LA6707-9				
						Heteroplasmic	LA6703-8				
						Homoplasmic	LA6704-6				
	6.1.12 Genomic Source Class	48002-0	LOINC	Code	VS	Germline	LA6683-2	LOINC	Observation.component:genomic-source-class	n/a	The classification of the genomic source, such as germline, somatic, or other origins.
						Somatic	LA6684-0				
						Fetal	LA10429-1				
						Likely germline	LA18194-3				
						Likely somatic	LA18195-0				
						Likely fetal	LA18196-8				
						Unknown genomic origin	LA18197-6				
						De novo	LA26807-0				
	6.1.13 DNA Change Type	48019-4	LOINC	Code	VS	Point Mutation	1000008	SO	Observation.component:coding-change-type	n/a	The variant's type of DNA change, such as point mutation, deletion, insertion, or other types.
						Deletion	159				
						Insertion	667				
						Insertion and Deletion	10000321				
						Substitution	1000002				
						Transition	1000009				
						Transversion	1000017				
						Trinucleotide-repeat-mutation	2165				
	6.1.14 Clinical Significance [ACMG]	53037-8	LOINC	Code	VSe	Pathogenic	LA6668-3	LOINC	Observation.component:clinical-significance Rec. VS: ClinicalSignificance	Interpretation. AcmgPathogenicityClassification	The clinical significance of the genetic variant, indicating its impact on health and disease.
						Likely pathogenic	LA26332-9				
						Uncertain significance	LA26333-7				
						Likely benign	LA26334-5				
						Benign	LA6675-8				
						Unknown	LA4489-6				
	6.1.15 Therapeutic Actionability	therapeutic_actionability	GA4GH	Code	VS	There is not enough information at this time to support any therapeutic actionability for this variant	UNKNOWN_ACTIONABILITY	GA4GH	n/a	[...].GenomicInterpretation. VariantInterpretation. therapeutic_actionability	An enumeration flagging the variant as being a candidate for treatment/ clinical intervention of the disorder caused by this variant, which could improve the clinical outcome.
						This variant has no therapeutic actionability.	NOT_ACTIONABLE				
						This variant is known to be therapeutically actionable.	ACTIONABLE				
	6.1.16 Clinical Annotation					Very strong evidence pathogenic	LA30200-2				The level of evidence supporting the clinical
						Strong evidence pathogenic	LA30201-0				
						Moderate evidence pathogenic	LA30202-8				
						Supporting evidence pathogenic	LA30203-6				

	Annotation Level Of Evidence	93044-6	LOINC	Code	VS	Supporting evidence benign	LA30204-4	LOINC	Observation.component:evidence-level	n/a	The level of evidence supporting the clinical annotation of the genetic variant.
						Strong evidence benign	LA30205-1				
						Stand-alone evidence pathogenic	LA30206-9				
						Stand-alone evidence benign	LA30207-7				
						Uncertain significance	LA26333-7				
6.2 Phenotypic Feature	**6.2.1 Phenotypic Feature**	8116006	SCT	Code	HPO	n/a			Observation.Code	PhenotypicFeature.type	An observed physical and clinical characteristic encoded with HPO.
	6.2.2 Determination Date	phenotypicfeature.onset	GA4GH	Date	YYYY-MM-DD	n/a			Observation.effectiveDateTime	PhenotypicFeature.onset	The date on which the phenotypic feature was observed or recorded. We recommend capturing the time a characteristic was observed.
	6.2.3 Status	phenotypicfeature.excluded	GA4GH	Code	VSe	Confirmed present	410605003	SCT	Observation.Status Rec VS: observation.status	PhenotypicFeature.excluded	The current status of the phenotypic feature, indicating whether it is confirmed or refuted.
						Refuted	723511001				
	6.2.4 Modifier	phenotypicfeature.modifier	GA4GH	Code	OntologyClass (e.g. HPO, NCBITAXON, SCT, etc.)	n/a			n/a	PhenotypicFeature.modifier	Any number of additional modifiers describing a specific phenotypic feature further, such as severity (HP:0012824), clinical modifiers (HP:0012823), or linking causative infectious agents using the NCBITAXON Ontology
	6.3.1 Propositus/-a	64245008	SCT	Code	VSe	Yes	373066001	SCT	n/a	(Family.relatives → 1 Phenopacket per family member)	Is the individual the first affected family member who seeks medical attention for a genetic disorder, leading to the diagnosis of other family members. Disclaimer: The SCT code for propositus (64245008) refers to any gender.
						No	373067005				
						Unknown	261665006				
						Not recorded	1220561009				
	6.3.2 Relationship of the individual to the index case / propositus/a	relationship_to_indexcase	SCT	Code	VSe	Natural mother	65656005	SCT	n/a Rec. VS: FamilyMember	(Family.relatives → 1 Phenopacket per family member)	Specifies the familial relationship of the individual being evaluated to the index case or propositus/proposita. Disclaimer: The SCT code for propositus (64245008) refers to any gender.
						Natural father	9947008				
						Natural daughter	83420006				
						Natural son	113160008				
						Natural brother	60614009				
						Natural sister	73678001				
						Twin sibling	11286003				
						Half-brother	45929001				
						Half-sister	2272004				
						Natural grandfather	62296006				
						Natural grandmother	17945006				
						Not recorded	1220561009				
	6.3.3 Consanguinity	842009	SCT	Code	VSe	Yes	373066001	SCT	n/a	Family.consanguinous_parents	The presence of a biological relationship between parents who are related by blood, typically as first or second cousins.
						No	373067005				
						Unknown	261665006				
						Not recorded	1220561009				
	6.3.4 Family		SCT	Code	VSe	Natural mother	65656005	SCT	FamilyMemberHistory.relationship.coding	Family.Pedigree.Person.	Specifies the relationship of the selected family
						Natural father	9947008				
						Natural daughter	83420006				
						Natural son	113160008				
						Natural brother	60614009				
						Natural sister	73678001				

6.3 Family History	***Member Relationship***	303071001	SCT	Code	VSe	Twin sibling	11286003	SCT	We recommend Use Rec. VS: FamilyMember	individual_id	Specifies the relationship of the selected family member to the patient.
						Half-brother	45929001				
						Half-sister	2272004				
						Natural grandfather	62296006				
						Natural grandmother	17945006				
						Not recorded	1220561009				
	6.3.5 Family Member Status	familymemberhistory.status	HL7 FHIR	Code	VS	Partial	partial	HL7 FHIR	FamilyMemberHistory.status	(Family.relatives)	Specifies the record's status of the family history of a specific family member.
						Completed	completed				
						Entered in Error	entered-in-error				
						Health Unknown	health-unknown				
	6.3.6 Family Member Sex	54123-5	LOINC	Code	VSe	Female	248152002	SCT	FamilyMemberHistory.sex Rec. VS: AdministrativeGender	Family.Pedigree.Person.sex	Specifies the sex (or gender) of the specific family member. If possible, the sex assigned at birth should be selected.
						Male	248153007				
						Gender unknown	394743007				
						Identifies as nonbinary gender	33791000087105				
						Not recorded	1220561009				
	6.3.7 Family Member Age	54141-7	LOINC	Integer	XXX	n/a			FamilyMemberHistory.age.value	(Family.relatives)	Records the current age of the selected family member.
	6.3.8 Family Member Date of Birth	54124-3	LOINC	Date	YYYY-MM-DD	n/a			FamilyMemberHistory.born.bornDate	(Family.relatives)	Records the date of birth of the selected family member.
	6.3.9 Family Member Deceased	740604001	SCT	Code	VSe	Yes	373066001	SCT	FamilyMemberHistory.deceased.deceasedBoolean	(Family.relatives)	Indicates whether the selected family member is deceased.
						No	373067005				
						Unknown	261665006				
	6.3.10 Family Member Cause of Death	54112-8	LOINC	Code	ICD10	n/a			FamilyMemberHistory.reasonCode	(Family.relatives)	Records the cause of death of the selected deceased family member.
	6.3.11 Family Member Deceased Age	54113-6	LOINC	Integer	XXX	n/a			FamilyMemberHistory.deceased.deceasedAge	(Family.relatives)	Records the age at which the selected family member died.
	6.3.12 Family Member Disease	familymemberhistory.condition	HL7 FHIR	Code	ORDO	n/a			FamilyMemberHistory.condition.code	(Family.relatives)	Indicates whether the selected family member is affected by the same RD, as the individual.
					ICD-10						
					ICD-11						
					MONDO						
					OMIM_g						
					OMIM_p						
	7.1 Consent Status	309370004	SCT	Code	VS	Pending	draft	HL7 FHIR	Consent.status	n/a	Indicates the current status of the consent.
						Proposed	proposed				
						Active	active				
						Rejected	rejected				
						Inactive	inactive				
						Entered in Error	entered-in-error				
	7.2 Consent Date	consent.datetime	SCT	Date	YYYY-MM-DD	n/a			Consent.dateTime	n/a	Records the date when the consent was given.

7. Consent	***7.3 Health Policy Monitoring***	386318002	SCT	String	n/a	n/a			Consent.policy	n/a	The references to the policies that are included in this consent scope. Policies may be organisational, but are often defined jurisdictionally, or in law.
	7.4 Agreement to be contacted for research purposes	LP192126-3	LOINC	Code	VSe	Yes	373066001	SCT	Consent.scope.coding	n/a	Indicates whether the patient agrees to be contacted for research purposes.
						No	373067005				
						Unknown	261665006				
	7.5 Consent to the reuse of data	413909005	SCT	Code	VSe	Yes	373066001	SCT	Consent.scope.coding	n/a	Indicates whether the patient consents to the reuse of their data.
						No	373067005				
						Unknown	261665006				
	7.6 Biological sample	123038009	SCT	Code	VSe	Yes	373066001	SCT	n/a	n/a	Indicates whether the patient consents to providing a biological sample.
						No	373067005				
						Unknown	261665006				
	7.7 Link to a biobank	840566006	SCT	String	n/a	n/a			n/a	n/a	If there is a biological sample, this data element indicates the link to the biobank of the patient's biological sample.
8. Disability	**8.1 Classification of functioning / disability**	icf_score	SCT	Code	ICF	n/a			n/a	n/a	Specifies the classification of the patient's functioning or disability according to the International Classification of Functioning, Disability and Health (ICF).